

Monique M Ryan

List of Publications by Year in descending order

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145
papers

6,028
citations

87888

38
h-index

85541

71
g-index

147
all docs

147
docs citations

147
times ranked

8063
citing authors

#	ARTICLE	IF	CITATIONS
1	Nusinersen initiated in infants during the presymptomatic stage of spinal muscular atrophy: Interim efficacy and safety results from the Phase 2 NURTURE study. <i>Neuromuscular Disorders</i> , 2019, 29, 842-856.	0.6	401
2	Ataluren in patients with nonsense mutation Duchenne muscular dystrophy (ACT DMD): a multicentre, randomised, double-blind, placebo-controlled, phase 3 trial. <i>Lancet, The</i> , 2017, 390, 1489-1498.	13.7	365
3	Ataluren treatment of patients with nonsense mutation dystrophinopathy. <i>Muscle and Nerve</i> , 2014, 50, 477-487.	2.2	357
4	A prospective evaluation of whole-exome sequencing as a first-tier molecular test in infants with suspected monogenic disorders. <i>Genetics in Medicine</i> , 2016, 18, 1090-1096.	2.4	332
5	Long-term effects of glucocorticoids on function, quality of life, and survival in patients with Duchenne muscular dystrophy: a prospective cohort study. <i>Lancet, The</i> , 2018, 391, 451-461.	13.7	306
6	Tyrosine hydroxylase deficiency: a treatable disorder of brain catecholamine biosynthesis. <i>Brain</i> , 2010, 133, 1810-1822.	7.6	268
7	Mutations in the RNA exosome component gene EXOSC3 cause pontocerebellar hypoplasia and spinal motor neuron degeneration. <i>Nature Genetics</i> , 2012, 44, 704-708.	21.4	216
8	Neurofilament as a potential biomarker for spinal muscular atrophy. <i>Annals of Clinical and Translational Neurology</i> , 2019, 6, 932-944.	3.7	137
9	Ascorbic acid for Charcot-Marie-Tooth disease type 1A in children: a randomised, double-blind, placebo-controlled, safety and efficacy trial. <i>Lancet Neurology, The</i> , 2009, 8, 537-544.	10.2	131
10	Pathophysiological Mechanisms of Dominant and Recessive GLRA1 Mutations in Hyperekplexia. <i>Journal of Neuroscience</i> , 2010, 30, 9612-9620.	3.6	112
11	<i>EPG5</i>-related Vici syndrome: a paradigm of neurodevelopmental disorders with defective autophagy. <i>Brain</i> , 2016, 139, 765-781.	7.6	99
12	Evolution of foot and ankle manifestations in children with CMT1A. <i>Muscle and Nerve</i> , 2009, 39, 158-166.	2.2	96
13	Next generation sequencing in a large cohort of patients presenting with neuromuscular disease before or at birth. <i>Orphanet Journal of Rare Diseases</i> , 2015, 10, 148.	2.7	94
14	Congenital Titinopathy: Comprehensive characterization and pathogenic insights. <i>Annals of Neurology</i> , 2018, 83, 1105-1124.	5.3	93
15	An open-label trial in Friedreich ataxia suggests clinical benefit with high-dose resveratrol, without effect on frataxin levels. <i>Journal of Neurology</i> , 2015, 262, 1344-1353.	3.6	89
16	Childhood chronic inflammatory demyelinating polyneuropathy: clinical course and long-term outcome. <i>Neuromuscular Disorders</i> , 2000, 10, 398-406.	0.6	87
17	Natural history of pulmonary function in collagen VI-related myopathies. <i>Brain</i> , 2013, 136, 3625-3633.	7.6	85
18	Pontocerebellar hypoplasia type 1. <i>Neurology</i> , 2013, 80, 438-446.	1.1	84

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19	Histopathological Findings in Hereditary Motor and Sensory Neuropathy of Axonal Type With Onset in Early Childhood Associated With <i>Mitofusin 2</i> Mutations. <i>Journal of Neuropathology and Experimental Neurology</i> , 2008, 67, 1097-1102.	1.7	81
20	Critical illness polyneuropathy and myopathy in pediatric intensive care: A review. <i>Pediatric Critical Care Medicine</i> , 2007, 8, 18-22.	0.5	75
21	Loss of function of SLC25A46 causes lethal congenital pontocerebellar hypoplasia. <i>Brain</i> , 2016, 139, 2877-2890.	7.6	74
22	Phase IIa trial in Duchenne muscular dystrophy shows vamorolone is a first-in-class dissociative steroidal anti-inflammatory drug. <i>Pharmacological Research</i> , 2018, 136, 140-150.	7.1	69
23	Guillain-Barre syndrome in childhood. <i>Journal of Paediatrics and Child Health</i> , 2005, 41, 237-241.	0.8	66
24	A new locus for X-linked dominant Charcot-Marie-Tooth disease (CMTX6) is caused by mutations in the pyruvate dehydrogenase kinase isoenzyme 3 (PDK3) gene. <i>Human Molecular Genetics</i> , 2013, 22, 1404-1416.	2.9	64
25	Vamorolone trial in Duchenne muscular dystrophy shows dose-related improvement of muscle function. <i>Neurology</i> , 2019, 93, e1312-e1323.	1.1	64
26	Auditory function in children with Charcot-Marie-Tooth disease. <i>Brain</i> , 2012, 135, 1412-1422.	7.6	63
27	Dietary L-Tyrosine Supplementation in Nemaline Myopathy. <i>Journal of Child Neurology</i> , 2008, 23, 609-613.	1.4	58
28	De Novo Truncating Mutations in AHDC1 in Individuals with Syndromic Expressive Language Delay, Hypotonia, and Sleep Apnea. <i>American Journal of Human Genetics</i> , 2014, 94, 784-789.	6.2	57
29	Health-related Quality of Life in Boys With Duchenne Muscular Dystrophy: Agreement Between Parents and Their Sons. <i>Journal of Child Neurology</i> , 2010, 25, 1188-1194.	1.4	55
30	Pediatric Guillain-Barré syndrome. <i>Current Opinion in Pediatrics</i> , 2013, 25, 689-693.	2.0	53
31	Neurophysiologic abnormalities in children with Charcot-Marie-Tooth disease type 1A. <i>Journal of the Peripheral Nervous System</i> , 2008, 13, 236-241.	3.1	49
32	Biomarkers and the Development of a Personalized Medicine Approach in Spinal Muscular Atrophy. <i>Frontiers in Neurology</i> , 2019, 10, 898.	2.4	49
33	NEMF mutations that impair ribosome-associated quality control are associated with neuromuscular disease. <i>Nature Communications</i> , 2020, 11, 4625.	12.8	47
34	Acute Transverse Myelitis and Acute Disseminated Encephalomyelitis in Childhood: Spectrum or Separate Entities?. <i>Journal of Child Neurology</i> , 2009, 24, 287-296.	1.4	46
35	Nusinersen for SMA: expanded access programme. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2018, 89, 937-942.	1.9	46
36	Hand involvement in children with Charcot-Marie-Tooth disease type 1A. <i>Neuromuscular Disorders</i> , 2008, 18, 970-973.	0.6	44

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37	The use of invasive ventilation is appropriate in children with genetically proven spinal muscular atrophy type 1: the motion against. Paediatric Respiratory Reviews, 2008, 9, 51-54.	1.8	44
38	Peripheral nerve ultrasound in pediatric Charcot-Marie-Tooth disease type 1A. Neurology, 2015, 84, 569-574.	1.1	42
39	Efficacy and safety of vamorolone in Duchenne muscular dystrophy: An 18-month interim analysis of a non-randomized open-label extension study. PLoS Medicine, 2020, 17, e1003222.	8.4	41
40	Meta-analyses of ataluren randomized controlled trials in nonsense mutation Duchenne muscular dystrophy. Journal of Comparative Effectiveness Research, 2020, 9, 973-984.	1.4	41
41	A mixed methods study of age at diagnosis and diagnostic odyssey for Duchenne muscular dystrophy. European Journal of Human Genetics, 2015, 23, 1294-1300.	2.8	39
42	Reliability of quantifying foot and ankle muscle strength in very young children. Muscle and Nerve, 2008, 37, 626-631.	2.2	36
43	Diagnostic and cost utility of whole exome sequencing in peripheral neuropathy. Annals of Clinical and Translational Neurology, 2017, 4, 318-325.	3.7	36
44	Effect of Oral Curcumin on DÃ©jÃ©rine-Sottas Disease. Pediatric Neurology, 2009, 41, 305-308.	2.1	35
45	Describing nutrition in spinal muscular atrophy: A systematic review. Neuromuscular Disorders, 2016, 26, 395-404.	0.6	35
46	â€A short time but a lovely little short timeâ€: Bereaved parents' experiences of having a child with spinal muscular atrophy type 1. Journal of Paediatrics and Child Health, 2016, 52, 40-46.	0.8	32
47	Onasemnogene abeparvovec in spinal muscular atrophy: an Australian experience of safety and efficacy. Annals of Clinical and Translational Neurology, 2022, 9, 339-350.	3.7	32
48	CMTX mimicking childhood chronic inflammatory demyelinating neuropathy with tremor. Muscle and Nerve, 2005, 31, 528-530.	2.2	31
49	Paralysis and a perihilar protuberance: An unusual presentation of sarcoidosis in a child. Pediatric Pulmonology, 2009, 44, 410-414.	2.0	31
50	MCM3AP in recessive Charcot-Marie-Tooth neuropathy and mild intellectual disability. Brain, 2017, 140, 2093-2103.	7.6	31
51	The severe epilepsy syndromes of infancy: A populationâ€based study. Epilepsia, 2021, 62, 358-370.	5.1	31
52	Spinal muscular atrophy type 1: Is long-term mechanical ventilation ethical?. Journal of Paediatrics and Child Health, 2007, 43, 237-242.	0.8	30
53	Physician attitudes towards ventilatory support for spinal muscular atrophy type 1 in Australasia. Journal of Paediatrics and Child Health, 2007, 43, 790-794.	0.8	30
54	Mutations in the cyclic adenosine monophosphate response element of the tyrosine hydroxylase gene. Annals of Neurology, 2007, 62, 422-426.	5.3	29

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55	Complete callosal agenesis, pontocerebellar hypoplasia, and axonal neuropathy due to AMPD2 loss. <i>Neurology: Genetics</i> , 2015, 1, e16.	1.9	29
56	A diagnostic approach to recurrent myalgia and rhabdomyolysis in children. <i>Archives of Disease in Childhood</i> , 2015, 100, 793-797.	1.9	29
57	Anterior horn cell disease and olivopontocerebellar hypoplasia. <i>Pediatric Neurology</i> , 2000, 23, 180-184.	2.1	28
58	Health status of boys with Duchenne muscular dystrophy: A parent's perspective. <i>Journal of Paediatrics and Child Health</i> , 2011, 47, 557-562.	0.8	27
59	Genetic axonal neuropathies and neuronopathies of pre-natal and infantile onset. <i>Journal of the Peripheral Nervous System</i> , 2012, 17, 285-300.	3.1	26
60	Interventions for the prevention and treatment of pes cavus. <i>The Cochrane Library</i> , 2007, , CD006154.	2.8	25
61	Development and validation of the Charcot-Marie-Tooth Disease Infant Scale. <i>Brain</i> , 2018, 141, 3319-3330.	7.6	25
62	Randomized trial of botulinum toxin to prevent pes cavus progression in pediatric charcot-marie-tooth disease type 1A. <i>Muscle and Nerve</i> , 2010, 42, 262-267.	2.2	24
63	Genetic, Radiologic, and Clinical Variability in Brown-Vialetto-van Laere Syndrome. <i>Seminars in Pediatric Neurology</i> , 2018, 26, 2-9.	2.0	24
64	Strong Correlation Between the 6-Minute Walk Test and Accelerometry Functional Outcomes in Boys With Duchenne Muscular Dystrophy. <i>Journal of Child Neurology</i> , 2015, 30, 357-363.	1.4	22
65	Quality of Life in Children With Charcot-Marie-Tooth Disease. <i>Journal of Child Neurology</i> , 2010, 25, 343-347.	1.4	21
66	Unraveling the pathogenesis of <i>ARX</i> polyalanine tract variants using a clinical and molecular interfacing approach. <i>Molecular Genetics & Genomic Medicine</i> , 2015, 3, 203-214.	1.2	21
67	DCC Is Required for the Development of Nociceptive Topognosis in Mice and Humans. <i>Cell Reports</i> , 2018, 22, 1105-1114.	6.4	21
68	Whole Genome Sequencing Identifies a 78 kb Insertion from Chromosome 8 as the Cause of Charcot-Marie-Tooth Neuropathy CMTX3. <i>PLoS Genetics</i> , 2016, 12, e1006177.	3.5	20
69	Childhood Chronic Inflammatory Demyelinating Polyneuropathy. <i>Journal of Child Neurology</i> , 2014, 29, 43-48.	1.4	19
70	Deterioration in gait and functional ambulation in children and adolescents with Charcot-Marie-Tooth disease over 12 months. <i>Neuromuscular Disorders</i> , 2017, 27, 658-666.	0.6	19
71	Importance of muscle biopsy to establish pathogenicity of DMD missense and splice variants. <i>Neuromuscular Disorders</i> , 2019, 29, 913-919.	0.6	19
72	Cardiac phenotype in <i>ATP1A3</i> -related syndromes. <i>Neurology</i> , 2020, 95, e2866-e2879.	1.1	19

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73	Neuronal ceroid lipofuscinosis type 2: an Australian case series. <i>Journal of Paediatrics and Child Health</i> , 2020, 56, 1210-1218.	0.8	19
74	High resolution chromosomal microarray in undiagnosed neurological disorders. <i>Journal of Paediatrics and Child Health</i> , 2013, 49, 716-724.	0.8	18
75	A multinational study on motor function in early-onset FSHD. <i>Neurology</i> , 2018, 90, e1333-e1338.	1.1	17
76	International retrospective natural history study of <i>LMNA</i>-related congenital muscular dystrophy. <i>Brain Communications</i> , 2021, 3, fcab075.	3.3	17
77	Epidermolysis bullosa with late-onset muscular dystrophy and plectin deficiency. <i>Muscle and Nerve</i> , 2011, 44, 135-141.	2.2	16
78	Autoimmune myasthenia gravis, immunotherapy and thymectomy in children. <i>Neuromuscular Disorders</i> , 2012, 22, 118-121.	0.6	16
79	Powered standing wheelchairs promote independence, health and community involvement in adolescents with Duchenne muscular dystrophy. <i>Neuromuscular Disorders</i> , 2019, 29, 221-230.	0.6	16
80	A homozygous <i>UBA5</i> pathogenic variant causes a fatal congenital neuropathy. <i>Journal of Medical Genetics</i> , 2020, 57, 835-842.	3.2	16
81	Current Therapeutic Strategies for Patients With Polyneuropathies Secondary to Inherited Metabolic Disorders. <i>Mayo Clinic Proceedings</i> , 2003, 78, 858-868.	3.0	15
82	Energy metabolism and mitochondrial defects in X-linked Charcot-Marie-Tooth (CMTX6) iPSC-derived motor neurons with the p.R158H PDK3 mutation. <i>Scientific Reports</i> , 2020, 10, 9262.	3.3	15
83	Atypical Silver-Russell phenotype resulting from maternal uniparental disomy of chromosome 7. <i>American Journal of Medical Genetics, Part A</i> , 2010, 152A, 2342-2345.	1.2	14
84	Demyelinating prenatal and infantile developmental neuropathies. <i>Journal of the Peripheral Nervous System</i> , 2012, 17, 32-52.	3.1	14
85	Recessive MYH7-related myopathy in two families. <i>Neuromuscular Disorders</i> , 2019, 29, 456-467.	0.6	14
86	Feasibility of a Computerized Method to Measure Quality of "Everyday" Life in Children with Neuromuscular Disorders. <i>Physical and Occupational Therapy in Pediatrics</i> , 2010, 30, 43-53.	1.3	13
87	Neurophysiological profile of peripheral neuropathy associated with childhood mitochondrial disease. <i>Mitochondrion</i> , 2016, 30, 162-167.	3.4	13
88	Gait and footwear in children and adolescents with Charcot-Marie-Tooth disease: A cross-sectional, case-controlled study. <i>Gait and Posture</i> , 2018, 62, 262-267.	1.4	13
89	A Randomized, Double-Blind, Placebo-Controlled, Global Phase 3 Study of Edasalonexent in Pediatric Patients with Duchenne Muscular Dystrophy: Results of the PolarisDMD Trial. <i>Journal of Neuromuscular Diseases</i> , 2021, 8, 769-784.	2.6	13
90	Juvenile polymyositis or paediatric muscular dystrophy: a detailed re-analysis of 13 cases. <i>Histopathology</i> , 2009, 55, 452-462.	2.9	12

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91	Pediatric sciatic neuropathy associated with neoplasms. <i>Muscle and Nerve</i> , 2011, 43, 183-188.	2.2	12
92	X-linked Recessive Distal Myopathy With Hypertrophic Cardiomyopathy Caused by a Novel Mutation in the FHL1 Gene. <i>Journal of Child Neurology</i> , 2015, 30, 1211-1217.	1.4	12
93	Pathogenic mechanisms underlying X-linked Charcot-Marie-Tooth neuropathy (CMTX6) in patients with a pyruvate dehydrogenase kinase 3 mutation. <i>Neurobiology of Disease</i> , 2016, 94, 237-244.	4.4	12
94	242nd ENMC International Workshop: Diagnosis and management of juvenile myasthenia gravis Hoofddorp, the Netherlands, 1â€“3 March 2019. <i>Neuromuscular Disorders</i> , 2020, 30, 254-264.	0.6	12
95	Physical activity of children and adolescents with Charcot-Marie-Tooth neuropathies: A cross-sectional case-controlled study. <i>PLoS ONE</i> , 2019, 14, e0209628.	2.5	11
96	Muscle cramp in pediatric Charcot-Marie-Tooth disease type 1A. <i>Neurology</i> , 2011, 77, 2115-2118.	1.1	10
97	Physical activity and the use of standard and complementary therapies in Duchenne and Becker muscular dystrophies. <i>Journal of Pediatric Rehabilitation Medicine</i> , 2016, 9, 55-63.	0.5	10
98	Falls in paediatric Charcot-Marie-Tooth disease: a 6-month prospective cohort study. <i>Archives of Disease in Childhood</i> , 2019, 104, 535-540.	1.9	10
99	Benefits of powered standing wheelchair devices for adolescents with Duchenne muscular dystrophy in the first year of use. <i>Journal of Paediatrics and Child Health</i> , 2020, 56, 1419-1425.	0.8	10
100	Pathogenic deep intronic MTM1 variant activates a pseudo-exon encoding a nonsense codon resulting in severe X-linked myotubular myopathy. <i>European Journal of Human Genetics</i> , 2021, 29, 61-66.	2.8	10
101	Gaining consent for publication in difficult cases involving children. <i>BMJ: British Medical Journal</i> , 2008, 337, a1231-a1231.	2.3	10
102	Clinical practice guideline for the management of paediatric Charcot-Marie-Tooth disease. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2022, 93, 530-538.	1.9	10
103	Establishment of the Australasian paediatric Charcot-Marie-Tooth disease registry. <i>Neuromuscular Disorders</i> , 2007, 17, 349-350.	0.6	9
104	Mobile arm supports in Duchenne muscular dystrophy: a pilot study of user experience and outcomes. <i>Disability and Rehabilitation: Assistive Technology</i> , 2021, 16, 880-889.	2.2	9
105	Scientific rationale for a higher dose of nusinersen. <i>Annals of Clinical and Translational Neurology</i> , 2022, 9, 819-829.	3.7	9
106	Can in-the-moment diary methods measure health-related quality of life in Duchenne muscular dystrophy?. <i>Quality of Life Research</i> , 2017, 26, 1145-1152.	3.1	8
107	Atypical childhood chronic inflammatory demyelinating polyneuropathy. <i>Muscle and Nerve</i> , 2010, 42, 293-295.	2.2	7
108	Clinical practice considerations in facioscapulohumeral muscular dystrophy Sydney, Australia, 21 September 2015. <i>Neuromuscular Disorders</i> , 2016, 26, 462-471.	0.6	7

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109	Therapeutic Options to Improve Bone Health Outcomes in Duchenne Muscular Dystrophy: Zoledronic Acid and Pubertal Induction. <i>Journal of Paediatrics and Child Health</i> , 2017, 53, 1247-1248.	0.8	7
110	Dejerineâ€“Sottas disease in childhoodâ€“Genetic and sonographic heterogeneity. <i>Brain and Behavior</i> , 2018, 8, e00919.	2.2	7
111	Delivering multidisciplinary neuromuscular care for children via telehealth. <i>Muscle and Nerve</i> , 2022, 66, 31-38.	2.2	7
112	Respiration-Related Laryngeal Electromyography in Children with Bilateral Vocal Fold Paralysis. <i>Annals of Otolaryngology, Rhinology and Laryngology</i> , 2010, 119, 791-795.	1.1	5
113	Extended treatment of childhood Charcotâ€“Marieâ€“Tooth disease with highâ€“dose ascorbic acid. <i>Journal of the Peripheral Nervous System</i> , 2011, 16, 272-274.	3.1	5
114	Spontaneous Intracranial Hypotension in Childhood: A Case Report and Review of the Literature. <i>Journal of Child Neurology</i> , 2011, 26, 761-766.	1.4	5
115	Carpal Tunnel Syndrome Secondary to Ganglion Cyst in a Child. <i>Journal of Child Neurology</i> , 2011, 26, 630-633.	1.4	5
116	Neuromuscular complications of intensive care. <i>Handbook of Clinical Neurology</i> / Edited By P J Vinken and G W Bruyn, 2013, 113, 1481-1483.	1.8	5
117	Cerebral palsy is not a diagnosis: A case report of a novel atlastinâ€“1 mutation. <i>Journal of Paediatrics and Child Health</i> , 2016, 52, 669-671.	0.8	5
118	Investigation of the activation of the temporalis and masseter muscles in voluntary and spontaneous smile production. <i>Journal of Plastic, Reconstructive and Aesthetic Surgery</i> , 2018, 71, 1051-1057.	1.0	5
119	The effects of calf massage in boys with Duchenne muscular dystrophy: a prospective interventional study. <i>Disability and Rehabilitation</i> , 2021, 43, 3803-3809.	1.8	5
120	Biallelic Variants in PYROXD2 Cause a Severe Infantile Metabolic Disorder Affecting Mitochondrial Function. <i>International Journal of Molecular Sciences</i> , 2022, 23, 986.	4.1	5
121	Neurophysiologic findings in children presenting with pes cavus. <i>Journal of the Peripheral Nervous System</i> , 2010, 15, 238-240.	3.1	4
122	Evaluation of Serial Casting for Boys with Duchenne Muscular Dystrophy: A Case Report. <i>Physical and Occupational Therapy in Pediatrics</i> , 2018, 38, 88-96.	1.3	4
123	Peripheral nerve disease secondary to systemic conditions in children. <i>Therapeutic Advances in Neurological Disorders</i> , 2019, 12, 175628641986636.	3.5	4
124	Distinct effects on mRNA export factor GANP underlie neurological disease phenotypes and alter gene expression depending on intron content. <i>Human Molecular Genetics</i> , 2020, 29, 1426-1439.	2.9	4
125	Effect of a multicomponent nutritional supplement on functional outcomes for Duchenne muscular dystrophy: A randomized controlled trial. <i>Clinical Nutrition</i> , 2021, 40, 4702-4711.	5.0	4
126	Juvenile Parkinsonism. <i>Journal of Paediatrics and Child Health</i> , 2013, 49, 409-411.	0.8	3

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127	Unique clinical and neurophysiologic profile of a cohort of children with CMTX3. <i>Neurology</i> , 2018, 90, e1706-e1710.	1.1	3
128	Infantile-Onset Myelin Protein Zero-Related Demyelinating Neuropathy Presenting as an Upper Extremity Monoplegia. <i>Seminars in Pediatric Neurology</i> , 2018, 26, 52-55.	2.0	3
129	Communication about spinal muscular atrophy and genetic risk within families: An Australian pilot study. <i>Journal of Paediatrics and Child Health</i> , 2020, 56, 1263-1269.	0.8	3
130	Generating an iPSC line (with isogenic control) from the PBMCs of an ACTA1 (p.Gly148Asp) nemaline myopathy patient. <i>Stem Cell Research</i> , 2021, 54, 102429.	0.7	3
131	Guillain-Barré syndrome with optic neuritis. <i>Journal of Paediatrics and Child Health</i> , 2022, 58, 887-890.	0.8	3
132	Gene therapy for neuromuscular disorders: prospects and ethics. <i>Archives of Disease in Childhood</i> , 2022, 107, 421-426.	1.9	3
133	The association between dietary factors and body weight and composition in boys with Duchenne muscular dystrophy. <i>Journal of Human Nutrition and Dietetics</i> , 2022, 35, 804-815.	2.5	3
134	Overview of Pediatric Peripheral Neuropathies. , 2015, , 274-288.		2
135	Neurologic Melioidosis: Case Report of a Rare Cause of Acute Flaccid Paralysis. <i>Journal of Pediatrics</i> , 2016, 170, 319-321.	1.8	2
136	Cerebellar ataxia with normal intellect associated with a homozygous truncating variant in <i>CA8</i> . <i>Clinical Genetics</i> , 2020, 97, 516-520.	2.0	2
137	VENLAFAXINE INGESTION IN A 4-YEAR-OLD GIRL. <i>Journal of Paediatrics and Child Health</i> , 2012, 48, 1047-1048.	0.8	1
138	A family with 2 X-linked disorders: Charcot-Marie-Tooth disease and hemophilia A. <i>Muscle and Nerve</i> , 2012, 46, 454-455.	2.2	1
139	Mononeuropathies. , 2015, , 243-273.		0
140	Acute Polyneuropathies. , 2015, , 379-397.		0
141	Disorders of the Ocular Motor Cranial Nerves and Extraocular Muscles. , 2015, , 922-957.		0
142	Authors' Response to Commentary. <i>Journal of Pediatric Rehabilitation Medicine</i> , 2016, 9, 77.	0.5	0
143	Fifty years of paediatric neurology in Australasia. <i>Journal of Paediatrics and Child Health</i> , 2016, 52, 861-864.	0.8	0
144	Comment on: Paediatric Facial Paralysis: An overview and Insights into Management. <i>Journal of Paediatrics and Child Health</i> , 2021, 57, 1725-1725.	0.8	0

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145	Characterising gait in paediatric neuromuscular disorders: an observational study of spatio-temporal gait in a clinical cohort. Disability and Rehabilitation, 2021, , 1-7.	1.8	0